



LPL gene

lipoprotein lipase

Normal Function

The *LPL* gene provides instructions for making an enzyme called lipoprotein lipase. This enzyme is found primarily on the surface of cells that line tiny blood vessels (capillaries) within muscles and in fatty (adipose) tissue. Lipoprotein lipase plays a critical role in breaking down fat in the form of triglycerides, which are carried from various organs to the blood by molecules called lipoproteins.

Lipoprotein lipase breaks down triglycerides carried by two different types of lipoproteins, which bring fat to the bloodstream from different organs. Fat from the intestine, which is taken in from the diet, is transported to the bloodstream by lipoproteins called chylomicrons. Another type of lipoprotein called very low density lipoprotein (VLDL) carries triglycerides from the liver to the bloodstream. When lipoprotein lipase breaks down triglycerides, the fat molecules are used by the body as energy or stored in fatty tissue for later use.

Health Conditions Related to Genetic Changes

familial lipoprotein lipase deficiency

More than 220 mutations in the *LPL* gene have been found to cause familial lipoprotein lipase deficiency. This condition disrupts the normal breakdown of triglycerides in the body, resulting in an increase of these fats. The most common mutation in people of European ancestry replaces the protein building block (amino acid) glycine with the amino acid glutamic acid at position 188 in the enzyme (written as Gly188Glu or G188E). Mutations that cause familial lipoprotein lipase deficiency reduce or eliminate lipoprotein lipase activity, which prevents the enzyme from effectively breaking down triglycerides in the bloodstream. As a result, triglycerides attached to lipoproteins accumulate in the blood and tissues, leading to inflammation of the pancreas (pancreatitis), enlarged liver and spleen (hepatosplenomegaly), fatty deposits in the skin (eruptive xanthomas), and the other signs and symptoms of familial lipoprotein lipase deficiency.

other disorders

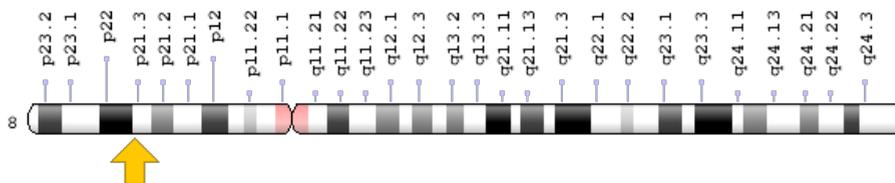
Certain variations in the *LPL* gene have been shown to influence the levels of fats in the bloodstream. The *LPL* gene variants likely result in the production of lipoprotein lipase enzymes with altered abilities to break down triglycerides. In some cases, the enzyme is overactive, resulting in low fat levels. In other cases, the enzyme

is impaired, resulting in increased fat levels, a condition called hyperlipidemia. Individuals with hyperlipidemia are at greater than normal risk of developing atherosclerosis, a condition in which fatty deposits accumulate on artery walls. This fatty material hardens over time, eventually blocking the arteries and increasing the chance of having a heart attack or stroke. It is unclear how much of a role *LPL* gene variants play in the development of atherosclerosis, as a large number of genetic and environmental factors determine the risk of developing this complex condition.

Chromosomal Location

Cytogenetic Location: 8p21.3, which is the short (p) arm of chromosome 8 at position 21.3

Molecular Location: base pairs 19,939,071 to 19,967,259 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- clearing factor lipase
- diacylglycerol lipase
- LIPD
- postheparin lipase
- triacylglycerol protein acylhydrolase

Additional Information & Resources

Educational Resources

- The Genetic Landscape of Diabetes: The Enzyme Lipoprotein Lipase
<https://www.ncbi.nlm.nih.gov/books/NBK1665/#A655>

GeneReviews

- Familial Lipoprotein Lipase Deficiency
<https://www.ncbi.nlm.nih.gov/books/NBK1308>

Genetic Testing Registry

- GTR: Genetic tests for LPL
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4023%5Bgeneid%5D>

Scientific articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LPL%5BTIAB%5D%29+OR+%28lipoprotein+lipase%5BTIAB%5D%29%29+AND+%28%28triacylglycero-protein+acylhydrolase%5BMAJR%5D%29+OR+%28lipemia-clearing+factor%5BMAJR%5D%29+OR+%28heparin-clearing+factor%5BMAJR%5D%29+OR+%28diglyceride+lipase%5BMAJR%5D%29+OR+%28postheparin+lipase%5BMAJR%5D%29+OR+%28post-heparin+lipase%5BMAJR%5D%29+OR+%28diacylglycerol+lipase%5BMAJR%5D%29+OR+%28postheparin+lipoprotein+lipase%5BMAJR%5D%29+OR+%28lipase,+lipoprotein%5BMAJR%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- HYPERLIPIDEMIA, FAMILIAL COMBINED
<http://omim.org/entry/144250>
- LIPOPROTEIN LIPASE
<http://omim.org/entry/609708>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_LPL.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=LPL%5Bgene%5D>
- HGNC Gene Family: Lipases
<http://www.genenames.org/cgi-bin/genefamilies/set/464>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6677
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4023>
- UniProt
<http://www.uniprot.org/uniprot/P06858>

Sources for This Summary

- Benlian P, De Gennes JL, Foubert L, Zhang H, Gagné SE, Hayden M. Premature atherosclerosis in patients with familial chylomicronemia caused by mutations in the lipoprotein lipase gene. *N Engl J Med*. 1996 Sep 19;335(12):848-54. Erratum in: *N Engl J Med* 1997 Feb 6;336(6):451.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/8778602>
- Gilbert B, Rouis M, Griglio S, de Lumley L, Laplaud P. Lipoprotein lipase (LPL) deficiency: a new patient homozygote for the preponderant mutation Gly188Glu in the human LPL gene and review of reported mutations: 75 % are clustered in exons 5 and 6. *Ann Genet*. 2001 Jan-Mar;44(1):25-32. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11334614>
- Kersten S. Physiological regulation of lipoprotein lipase. *Biochim Biophys Acta*. 2014 Jul;1841(7):919-33. doi: 10.1016/j.bbali.2014.03.013. Epub 2014 Apr 8. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24721265>
- Mead JR, Irvine SA, Ramji DP. Lipoprotein lipase: structure, function, regulation, and role in disease. *J Mol Med (Berl)*. 2002 Dec;80(12):753-69. Epub 2002 Oct 24. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12483461>
- Mead JR, Ramji DP. The pivotal role of lipoprotein lipase in atherosclerosis. *Cardiovasc Res*. 2002 Aug 1;55(2):261-9. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12123765>
- Pirim D, Wang X, Radwan ZH, Niemsiri V, Hokanson JE, Hamman RF, Barmada MM, Demirci FY, Kamboh MI. Lipoprotein lipase gene sequencing and plasma lipid profile. *J Lipid Res*. 2014 Jan; 55(1):85-93. doi: 10.1194/jlr.M043265. Epub 2013 Nov 9.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24212298>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3927465/>
- Tang W, Apostol G, Schreiner PJ, Jacobs DR Jr, Boerwinkle E, Fornage M. Associations of lipoprotein lipase gene polymorphisms with longitudinal plasma lipid trends in young adults: The Coronary Artery Risk Development in Young Adults (CARDIA) study. *Circ Cardiovasc Genet*. 2010 Apr;3(2):179-86. doi: 10.1161/CIRCGENETICS.109.913426. Epub 2010 Feb 11.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20150529>
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